International Journal of Research in Medical Sciences
Ghritlaharey RK et al. Int J Res Med Sci. 2023 Dec;11(12):xxx-xxx
www.msjonline.org
pISSN 2320-6071 | eISSN 2320-6012

DOI: https://dx.doi.org/10.18203/2320-6012.ijrms20233539

Case Series

Delayed presentation of congenital diaphragmatic hernia in children: case series and review of the literature

Rajendra K. Ghritlaharey*, Yogshri Chaubey

Department of Pediatric Surgery, Gandhi Medical College and Associated, Kamla Nehru and Hamidia Hospitals, Bhopal, Madhya Pradesh, India

Received: 22 October 2023
Accepted: 08 November 2023

***Correspondence:**
Dr. Rajendra K. Ghritlaharey,
E-mail: drrajendrak1@gmail.com

ABSTRACT

Congenital diaphragmatic hernia (CDH) is the most common developmental defect of the diaphragm. Ten to 20% of CDH cases may clinically present later in life, after the neonatal period. This study aims to review the demographics, clinical characteristics, various radiological investigations carried out for the diagnosis, and outcome of the surgical therapy offered for the treatment of late-presenting CDH in children. This study is a retrospective, descriptive study of the children diagnosed with CHD, clinically presented late after the neonatal period, and were surgically treated. These children were admitted and managed in the author's department of pediatric surgery between January 2019 and September 2023. During the study period, n=5 children (n=3 boys and n=2 girls) were diagnosed as delayed-presenting CDH and were operated upon by the authors. Their age ranged from two to ten months, with a mean of 5.6 months. Four children were diagnosed with left-sided CDH and one of the children with a right-sided CDH. All the children were operated upon through the abdominal incision. The reduction of the hernia contents into the peritoneal cavity and primary repair of the diaphragmatic defect was carried out successfully in all the children. The postoperative recovery was excellent for all the cases. All five children did well during the follow-up period, ranging from one month to four years. Children diagnosed with a delayed-presenting CDH, clinically presented with vague symptoms. Surgical therapy offered for the management of the delayed-presenting above-mentioned CDH cases carries an excellent result.

Keywords: Children, CDH, Delayed presentation, Diagnosis, Surgical therapy, Ectopic kidney

INTRODUCTION

Congenital diaphragmatic hernia (CDH) is a common developmental defect of the diaphragm.[1,2] The incidence of CDH is one in 2000 to 5000 live births.[2,3] CDH is more common on the left side and is reported to occur in approximately 80% of the cases.[2,4,5] Most CDH cases are clinically present within 24 hours of life.[2,3] Ten to 20% of CDH cases present clinically after the neonatal period and are called delayed/late-presenting CDH.[2,6] The clinical presentation of the delayed diagnosed CDH cases differs from that of the CDH cases presenting early in neonatal life.[2,6,7] Although in most cases a plain skiagram of the chest is suggestive of CHD, a computed tomography (CT) scan of the chest is also carried out/ordered for the confirmation of the diagnosis in many of the delayed-presenting CDH cases.[6,7] The surgical repair of late-presenting CDH cases can be accomplished either through the abdominal or thoracic route and, can be done either by open or using minimal access techniques.[2,6-8] This manuscript reports our experience of the management of n=5 children who were diagnosed as delayed-presenting CDH. A brief review of the literature is also included in this article.

CASE SERIES

This study is retrospectively carried out in children who were diagnosed as a case of delayed-presenting CDH.

These children were admitted and managed between January 2019 and September 2023 in the author's Department of Pediatric Surgery. The clinical records of the children mentioned above were reviewed in detail. Cases diagnosed and operated for CDH during the neonatal period were excluded. Unoperated delayed-presenting CDH cases, and cases with incomplete desired details were excluded. Consent was obtained from the parents of above-described children regarding the presentation/publication of clinical data, radiological investigations, and clinical and operative photographs of their child. Five children (n=3 boys and n=2 girls) were diagnosed with delayed-presenting CDH and were operated upon by the authors during the study period. Demographics, diagnostic modalities used, surgical therapy executed, postoperative complications, and outcomes of the surgical therapy offered for the above n=5 children are detailed in Table 1. Their age ranged from 2 to 10 months, with a mean of 5.6 months. Clinically all the children presented with vague respiratory symptoms, pneumonia, and mild other abdominal symptoms. Plain skiagram of the chest and abdomen (n=5), (Figure 1) ultrasonography (USG) of the chest and abdomen (n=5), and computed tomography (CT) scan of the chest and abdomen (n=4) were the radiological investigations obtained. CT scan of the chest and abdomen was obtained for n=4 cases and was diagnostic of CDH (Figure 2). CT scan of the chest and abdomen carried out on a boy also revealed the presence of an intrathoracic location of his right kidney, along with a right-sided CDH (Figure 3). Four children were diagnosed with a left-sided CDH and one of the children with a right-sided CDH. All the children were electively operated upon through the open abdominal incision. The contents of left-sided CDH were the part of the left lobe of the liver, stomach, the small intestine, part of the colon, and spleen (Figure 4A). One of the boys was diagnosed with right-sided CDH, contents were part of the right lobe of the liver, small intestine, part of the colon, and his right kidney was also ectopically located in the intrathoracic position (Figure 4B).

Table 1: Demographics, radiological diagnosis, operative procedures performed, and outcome of the cases of delayed presented CDH (n=5).

Month/ year operated	Age / sex (in months)	Radiological diagnosis	Size of defect	Operative procedure executed	Complication	Follow-up (Months)	Remark
July 2019	3, female	Left sided CDH	4 cm	Reduction of the contents and repair of diaphragmatic defect	Stitch granuloma	50	Well
Nov. 2021	2, male	Left sided CDH	4 cm	Reduction of the contents, excision of hernia sac, and repair of diaphragmatic defect	Nil	22	Well
April 2022	5, male	Left sided CDH	4.5 cm	Reduction of the contents, excision of the hernia sac, and repair of diaphragmatic defect	Nil	12	Well
May 2023	10, male	Right sided CDH	4.5 cm	Reduction of the contents, excision of the hernia sac, and repair of diaphragmatic defect	Nil	4	Well
Sept. 2023	8, female	Left sided CDH	5 cm	Reduction of the contents and repair of diaphragmatic defect	Nil	1	Well

All the children were operated upon through the abdominal incision. Left subcostal skin incision was preferred for left-sided CHD, and in the case of right-sided CDH, midline supra-umbilical incision was preferred. The reduction of the hernia contents into the peritoneal cavity and primary repair of the diaphragmatic defect was carried out successfully in all the children. In three cases, there was a hernia sac (Figure 5 A and B) that needed excision before the primary repair. In one boy, his right intrathoracic ectopic kidney was also brought down easily into the retroperitoneal area. The lung of the affected side was hypoplastic in all five children. The postoperative recovery was excellent for all the cases. All five children did well during the follow-up period, ranging from one month to four years. A telephonic conversation was done recently with the parents and all were happy and satisfied with the physiological and other development of their child.

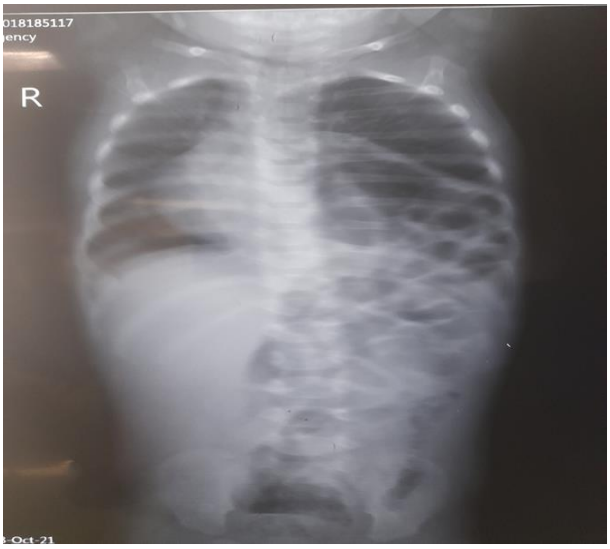

Figure 1: Plain skiagram of chest and abdomen showing continuity of bowel gas pattern into the left side of the chest, shift of the mediastinum towards the right side, suggestive of left-sided CDH (case 2).

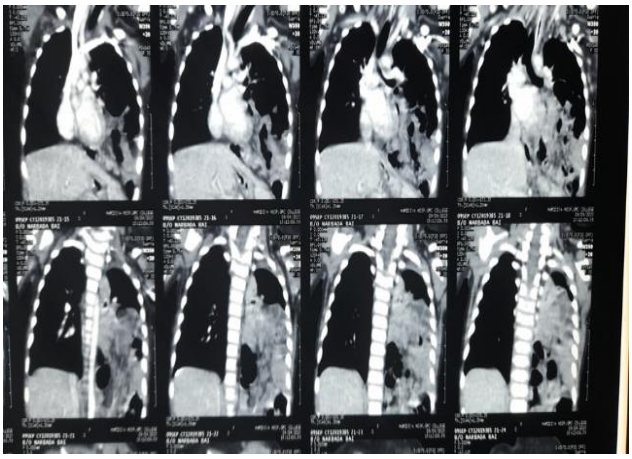

Figure 2: CT of chest and abdomen showing left-sided CDH (case 5).

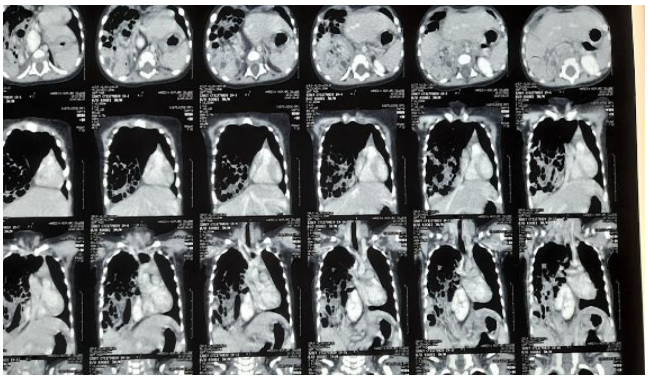

Figure 3: CT of chest and abdomen showing bowel loops within right side of chest, left normal positioned kidney, empty right kidney fossa area, and right ectopic intrathoracic kidney in 10 months-old-boy diagnosed right-sided CDH (case 4).

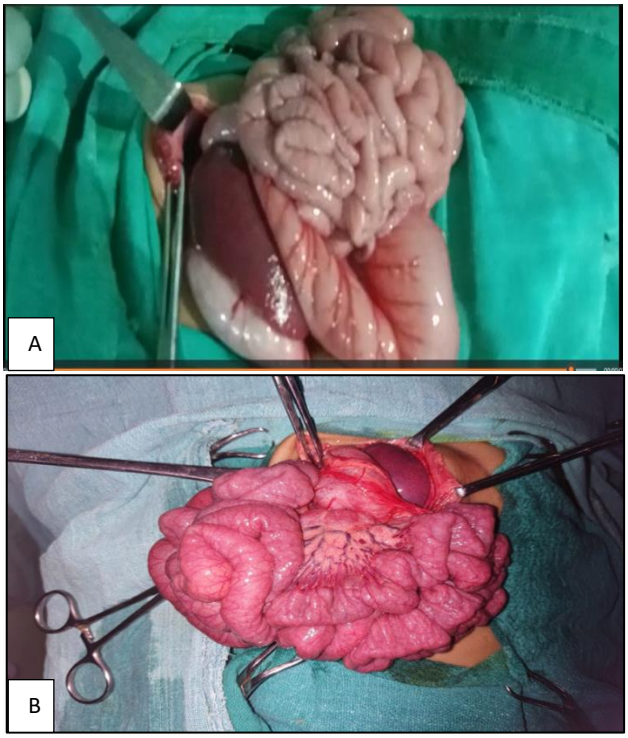

Figure 4 (A and B): Intra-op picture of contents (small intestine, part of colon, stomach, and spleen) in child operated upon for left-sided CDH (case 2), contents (small intestine, part of colon, and right kidney) in child operated upon for right-sided CDH (case 4).

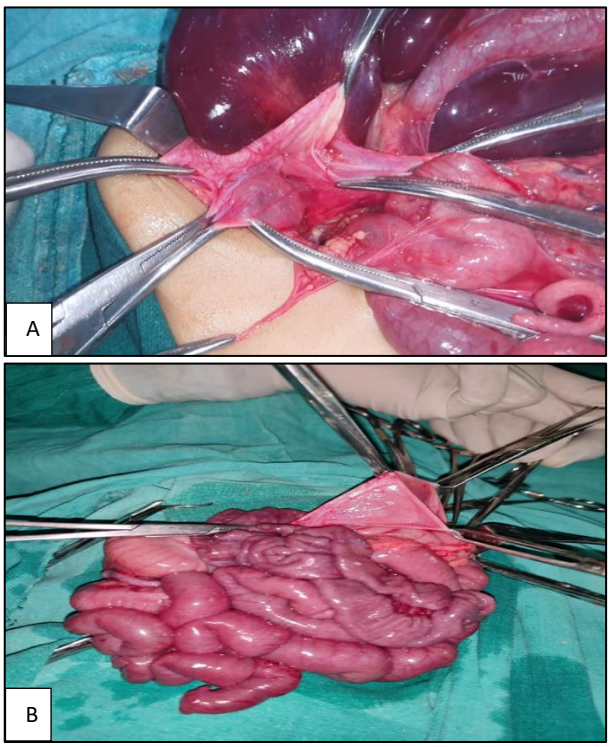

Figure 5 (A and B): Intra-op picture of diaphragmatic hernia sac in child operated for right-sided CDH (case 4) and diaphragmatic hernia sac in child operated for left-sided CDH (case 3)

DISCUSSION

The incidence of CDH is estimated to occur 1 in 2000 to 5000 births.[1,2,3,9] Due to the associated lethal congenital anomalies, approximately one-third of infants diagnosed with CDH are stillborn.[2,9] It occurs in the left side of the diaphragm in 80% of the cases.[1,2,4] In 10% to 20% of the cases, it occurs on the right side of the diaphragm.[2,4] Bilateral CDH is extremely rare, reported to occur in 1 to 2% of all CDH cases, and it is associated with very high mortality as well.[2,10] Associated congenital anomalies are reported in 10% to 50% of the infants diagnosed with CDH. Associated congenital anomalies are also more common in infants diagnosed with bilateral CDH and are associated with high mortality.[2,3,10] Although the exact causes of the CDH are not known, the most accepted theory is that the failure of the union of the parts of the developing diaphragm results in the CDH.[1,2] The classical left-sided CDH is a posterolateral defect in the diaphragm and is 2 to 4 cm. It is also known as Bochdalek's hernia and is reported in 80% to 90% of all CDH cases.[1,2,4]

In general, the spectrum of respiratory symptoms in infants born with CDH is determined by the degree/severity of the pulmonary hypoplasia, reactive pulmonary hypertension, and associated congenital anomalies. Most infants are clinically present with respiratory distress within the first 24 hours of their life.[2] The findings of respiratory distress, dextrocardia, and scaphoid abdomen are considered as CDH until proven otherwise. Ten to 20% of CDH cases may present later in life after the neonatal period and present with mild respiratory symptoms, pneumonia, and abdominal symptoms.[2,6-8] Few of the late-presenting CDH cases may present with features of respiratory failure, intestinal obstruction, and gastric volvulus.[2,6-8,11] Misdiagnosis as pneumothorax and pleural effusion has also been made for the delayed- presenting CDH in a few of the cases.[7,11]

In general, in up to 90% of the cases, the diagnosis of CDH is often made during the antenatal USG examination.[2,5,12] The mean gestational age at the diagnosis of CDH is 24 weeks, although reported as early as the gestational age of 11 weeks.[2] Antenatal USG is a gold standard diagnostic tool for the detection of CDH.[12] Fetal magnetic resonance imaging (MRI) plays a crucial role in better evaluation of the contents within the chest and fetal lungs in the USG-detected CDH and is required and also carried out with increasing frequency.[12,13]

The diagnosis of late/delayed-presenting CDH can be confirmed by a plain radiograph of the chest and abdomen demonstrating the presence of the bowel gas pattern within the chest. Insertion of a nasogastric tube prior to the skiagram of a chest and abdomen is also helpful for detecting gastric air bubbles within the chest cavity.[14,15] An angulation of the mediastinum and shifting of the cardiac shadow towards the contralateral side are also supportive findings on plain skiagram of the chest and abdomen.[6,11] Gastrointestinal dye studies/contrast studies are mostly not required for confirming the diagnosis of CDH, but obtained/needed for some of the cases.[6,14] CT scans of the chest and abdomen are frequently carried out before the surgical therapy offered for children diagnosed with a CDH and clinically presenting late after the neonatal period.[6,7,14] In the present series of n=5 cases, late-presenting CDH was confirmed on CT scans of the chest and abdomen in four children. CT scan of the chest and abdomen was helpful in exclusion of other congenital diseases viz eventration of the diaphragm, anterior diaphragmatic hernia (Morgagni's hernia), and other cystic lesions of the lung. CT scan of the chest and abdomen obtained in a boy, a right intrathoracic ectopic kidney was also detected along with a right-sided CDH.

Repair of the diaphragmatic defect of late-presenting CDH may be executed through one of the following techniques; (a) open laparotomy, (b) open thoracotomy, (c) laparoscopically, or (d) thoracoscopically using techniques of minimal access surgery (MIS).[6-8,11,16] Most surgeons prefer an open, subcostal abdominal approach for the surgical repair of the diaphragmatic defect of CDH.[6,7] The advent of MIS represents one of the most important surgical developments of the modern era and has seen significant growth and development over the past 30 years. The benefits of MIS for CDH cases operated during the early neonatal/neonatal period are well recognized.[17-20] The benefits of MIS for delayed-presenting CDH cases compared to traditional open surgery are also well recognized.[8,11,16] As a standard procedure during the surgical procedures executed for CDH cases, a primary repair using non-absorbable, interrupted sutures must be performed, if the diaphragmatic defect is adequate.[2,6,7,16,21] For the infants/children if the diaphragmatic defect is larger and a primary repair is not possible, in such cases, modifications/other reconstructive techniques are recommended, including mesh repair.[2,5-7,21]

The diaphragmatic defect of CDH is an open space between the chest and abdomen. There is a direct communication/anatomical communication between thoracic and abdominal cavities. In approximately 20% of CDH cases, a hernia sac is present. The hernia sac is a membrane of the parietal pleura and the peritoneum. The hernia sac must be excised before the repair of the diaphragmatic defect to minimize the recurrence. The presence of a hernia sac is reported to have better outcomes and lesser requirement of patch repair during surgical procedures carried out for CDH. [2,22,23] All five children of the present series were operated upon through the open techniques and via abdominal approach. A left subcostal incision was given for four children diagnosed with left-sided CDH, and a midline, supraumbilical incision was given for a boy diagnosed with right-sided CDH. Three children of the present series documented diaphragmatic hernia sac and required excision before repair of the diaphragmatic defect. Primary repair of diaphragmatic defect was achieved successfully in all

five children. Stretching of the abdominal wall was not required in our cases, and abdominal wall closure was achieved without compromising the intra-abdominal pressure. Additional surgical procedures were neither required nor carried out in any of the children. Prophylactically, an infant feeding tube was inserted for intercostal chest tube drainage in all five children, although it is not always required.

In general, the recurrence of the CDH is one of the known but unpredictable complications occurring after the successful repair of the CDH, and it depends upon various factors. The incidence of recurrent diaphragmatic hernia ranged from 10% to 25% of the cases.[5,18,21,24] Recurrent diaphragmatic hernia is more frequently documented and reported in children treated with patch repair. Most of the recurrent diaphragmatic hernia occurs on the left side. The most frequently recurrent diaphragmatic hernia occurred within the first 12 months to 24 months after the initial CDH repair and ranged from a few weeks to the first 5 years.[5,18,24] Most frequently, the surgical procedure for recurred CDH cases is approached through the abdominal route, but can also be done thoracoscopically. If adequate diaphragmatic tissues are available, primary repair of the defect must be attempted. For other cases, other options are available.[24,25] In the present study of five children, a primary repair was possible and achieved successfully. None of the children have a recurrence in the follow-up, ranging from one month to four years.

CONCLUSION

Children diagnosed as delayed-presenting CDH clinically presented with vague symptoms. In four-fifths of cases, thoracic and abdominal computed tomography scans were required for the confirmation of the diagnosis. Surgical therapy offered and executed through the abdominal route for the repair of the diaphragmatic defect in delayed-presenting above-mentioned CDH cases carries an excellent result.

Funding: No funding sources
Conflict of interest: None declared
Ethical approval: Not required

REFERENCES

1. Merrell AJ, Kardon G. Development of the diaphragm-a skeletal muscle essential for mammalian respiration. FEBS J. 2013;280:4026-35.
2. Charles JH, Stolar, Peter W. Dillon. Congenital diaphragmatic hernia and eventration. In: Pediatric Surgery. Coran AG, Adzick NS, Krummel TM, Laberge, editors. 7th ed. Philadelphia, PA: Elsevier Inc. Mosby. 2012;809-84.
3. Politis MD, Bermejo-Sánchez E, Canfield MA, Contiero P, Cragan JD, Dastgiri S et al. Prevalence and mortality in children with congenital diaphragmatic hernia: A multicountry analysis. Ann Epidemiol. 2021;56:61-9.e3.
4. Partridge EA, Peranteau WH, Herkert L, Rendon N, Smith H, Rintoul NE, et al. Right- versus left-sided congenital diaphragmatic hernia: a comparative outcomes analysis. J Pediatr Surg. 2016;51:900-2.
5. Gerall CD, Stewart LA, Price J, Kabagambe S, Sferra SR, Schmaedick MJ et al. Long-term outcomes of congenital diaphragmatic hernia: A single institution experience. J Pediatr Surg. 2022;57:563-9.
6. Cigdem MK, Onen A, Otcu S, Okur H. Late presentation of Bochdalek-type congenital diaphragmatic hernia in children: A 23-year experience at a single center. Surg Today. 2007;37:642-5.
7. Hamid R, Baba AA, Shera AH, Wani SA, Altaf T, Kant MH. Late-presenting congenital diaphragmatic hernia. Afr J Paediatr Surg. 2014;11:119-23.
8. Obata S, Souzaki R, Fukuta A, Esumi G, Nagata K, Matsuura T et al. Which is the better approach for late-presenting congenital diaphragmatic hernia: Laparoscopic or thoracoscopic? A single institution's experience of more than 10 years. J Laparoendosc Adv Surg Tech A. 2020;30:1029-35.
9. Balayla J, Abenhaim HA. Incidence, predictors and outcomes of congenital diaphragmatic hernia: a population-based study of 32 million births in the United States. J Matern Fetal Neonatal Med. 2014;27:1438-44.
10. Botden SM, Heiwegen K, van Rooij IA, Scharbatke H, Lally PA, van Heijst A et al. Bilateral congenital diaphragmatic hernia: prognostic evaluation of a large international cohort. J Pediatr Surg. 2017;52:1475-79.
11. Singh S, Wakhlu A, Pandey A, Kureel SN, Rawat JD. Delayed presentation of strangulated congenital diaphragmatic hernia: learning from our experience. Hernia. 2013;17:403-7.
12. Caro-Domínguez P, Victoria T, Ciet P, De la Torre E, Toscano ÁC, Diaz LG et al. Prenatal ultrasound, magnetic resonance imaging and therapeutic options for fetal thoracic anomalies: a pictorial essay. Pediatr Radiol. 2023;53:2106-19.
13. Amodeo I, Borzani I, Raffaeli G, Persico N, Amelio GS, Gulden S et al. The role of magnetic resonance imaging in the diagnosis and prognostic evaluation of fetuses with congenital diaphragmatic hernia. Eur J Pediatr. 2022;181:3243-57.
14. Bagłaj M, Dorobisz U. Late-presenting congenital diaphragmatic hernia in children: a literature review. Pediatr Radiol. 2005;35:478-88.
15. Rattan KN, Singh J, Dalal P. Diagnostic challenges in late-presenting congenital diaphragmatic hernia: a 16-year experience from tertiary care centre in North India. Trop Doct. 2019;49:138-41.
16. Yuan M, Li F, Xu C, Fan X, Xiang B, Huang L et al. Thoracoscopic treatment of late-presenting congenital diaphragmatic hernia in infants and

children. J Laparoendosc Adv Surg Tech A. 2019;29:77-81.
17. Vijfhuize S, Deden AC, Costerus SA, Sloots CE, Wijnen RM. Minimal access surgery for repair of congenital diaphragmatic hernia: is it advantageous? An open review. Eur J Pediatr Surg. 2012;22:364-73.
18. Lacher M, St Peter SD, Laje P, Harmon CM, Ure B, Kuebler JF. Thoracoscopic CDH repair--A survey on opinion and experience among IPEG members. J Laparoendosc Adv Surg Tech A. 2015;25:954-7.
19. Tyson AF, Sola R Jr, Arnold MR, Cosper GH, Schulman AM. Thoracoscopic versus open congenital diaphragmatic hernia repair: Single tertiary center review. J Laparoendosc Adv Surg Tech A. 2017;27:1209-16.
20. Okawada M, Ohfuji S, Yamoto M, Urushihara N, Terui K, Nagata K et al. Thoracoscopic repair of congenital diaphragmatic hernia in neonates: findings of a multicenter study in Japan. Surg Today. 2021;51:1694-702.
21. Heiwegen K, de Blaauw I, Botden SMBI. A systematic review and meta-analysis of surgical morbidity of primary versus patch repaired congenital diaphragmatic hernia patients. Sci Rep. 2021;11:12661.
22. Bouchghoul H, Marty O, Fouquet V, Cordier AG, Senat MV, Saada J et al. Congenital diaphragmatic hernia has a better prognosis when associated with a hernia sac. Prenat Diagn. 2018;38:638-44.
23. Raitio A, Salim A, Losty PD. Congenital diaphragmatic hernia-does the presence of a hernia sac improve outcome? A systematic review of published studies. Eur J Pediatr. 2021;180:333-7.
24. Słowik-Moczydłowska Ż, Kamiński A. Recurrent congenital diaphragmatic hernia: A single center experience. Pediatr Med Chir. 2021;43:228.
25. Gohda Y, Yokota K, Uchida H, Shirota C, Tainaka T, Sumida W et al. Safe thoracoscopic repair of recurrent congenital diaphragmatic hernia after initial open abdominal repair. Surg Today. 2023;37831144.

Cite this article as: Ghritlaharey RK, Chaubey Y. Delayed presentation of congenital diaphragmatic hernia in children: case series and review of the literature. Int J Res Med Sci 2023;11:xxx-xx.